

**State Plan  
For  
Genetic Services in Missouri**

# **Plan for Genetic Services in Missouri**

## **I. Introduction**

Genetic and genetic-related disorders have a tremendous impact on the health of the general population.

- Each year approximately 80,000 newborns are added to Missouri's population.
- At birth, over 2,000 of these infants will have a readily observable genetic disorder.
- An estimated 20% of all families are affected by a genetic or genetic related disorder.
- Chromosomal abnormalities occur in slightly more than 1 in 200 newborns and account for up to 60% of all miscarriages.
- It is estimated that genetic conditions contribute to 40% of all infant mortality.

Most diseases have a genetic component, whether inherited or resulting from the body's response to environmental factors. Knowledge of genetics is expanding rapidly, partly due to the Human Genome Project, which has mapped, and aims to analyze, every human gene. This progress in genetics will result in advanced technology that will increase the understanding of the pathophysiology of common diseases, opportunities to prevent diseases, earlier and more effective treatment, more effective therapies, and fewer side effects from therapies.

This plan supports the Missouri Department of Health and Senior Services' (DHSS, known as DOH – Department of Health prior to August 28, 2001) vision of Healthy Missourians for Life through the responsible use of genetic information. The overall goal of genetic services is to reduce morbidity and mortality associated with genetic disorders. The purpose of this document is to provide Missouri with future direction over the next three years, that optimizes the potential benefits of new developments and new opportunities to more effectively supply genetic services to residents. This report includes a demographic overview of the State, a description of genetic resources, assessment activities taken to identify barriers and gaps in

services, and goals and objectives to achieve the reduction of morbidity and mortality associated with genetic disorders.

## **II. Missouri Demographic Overview**

Illinois, Kentucky, Tennessee, Arkansas, Oklahoma, Kansas, Nebraska, and Iowa border the State of Missouri, located near the geographic center of the United States. Existing medical referral patterns suggest there is considerable crossing of state lines to receive genetic and other health care services.

A little over half (52.2 %) of Missouri's 5.8 million people are concentrated within the State's two major metropolitan areas, St. Louis and Kansas City. Another 10.5% of Missourians reside within the State's minor metropolitan areas of Springfield, Joplin, Columbia, and St Joseph. The rest are dispersed throughout the remaining 74 counties, 28 of which have fewer than 10,000 people.

As of July 1, 2007, there were 5,878,415 persons living in Missouri. The 2007 population estimates revealed that 98.62 percent of the population declared one race. Of that, 85.07 percent were white, 11.52 percent were African American, 0.49 percent were American Indian/Alaskan Native, 1.45 percent were Asians, 0.07 percent were Native Hawaiian/Pacific Islander and 1.37 percent were a combination of more than one race, or other. By ethnicity, 3.03 percent were of Hispanic or Latino origin. This is an increase from the 2000 census of .90 percent.

African Americans are still the dominant minority population in Missouri -- a total population of 677,657 in 2007. In 2007, African Americans accounted for 11.5 percent of Missouri's total population, up slightly from 10.8 percent of the State total population in 1990. The African American population is highly concentrated in the two major metropolitan areas, the City of St. Louis and Kansas City in Jackson County.

Since 1990, Hispanics have been the fastest growing minority group in Missouri. It is now estimated that there are 178,421 Hispanics or Latinos living in Missouri who

now make up approximately 3 percent of the total population. The Hispanic population resides primarily within the metropolitan areas. The Jewish community is centered in St. Louis and Kansas City. A significant Amish and Mennonite population is found in Central and North Central Missouri. Other genetically important ethnic groups, including Asians and those of Mediterranean descent, are numerically small and reside primarily in the metropolitan areas of the State.

The population of women of childbearing age is increasing. In 1995 there were 1,183,100 females between the ages of 15 and 44 in Missouri. By 2006 there were 1,197,935 in this same age group.

### **III. Genetic Services in Missouri**

Genetic activities in the DHSS date back to 1965 with the establishment of newborn screening (NBS) for phenylketonuria (PKU). Through the years the following tests were added to the NBS, congenital hypothyroid was added in 1979, galactosemia in 1985, sickle cell, and other hemoglobinopathies in 1989, congenital adrenal hyperplasia, and universal newborn hearing screening in 2002, expanded newborn screening using tandem mass technology in 2005, and cystic fibrosis (CF) in 2007.

In 1972 a statewide sickle cell screening, education, and counseling program was established with state funds. The development of reimbursement programs for the treatment of CF, Hemophilia, and Sickle Cell Anemia programs rapidly followed. In 1981, Missouri received federal funding to support the development of the Missouri Genetic Disease Program (MGDP), which consisted of a dynamic partnership between the DHSS and the four university affiliated medical centers in the State, to establish a network of genetic services. When federal funding ended in 1986, State legislation ([Attachment A](#)) was passed and State funding was established to support infrastructure at the four genetic centers to continue genetic activities. With the passage of this legislation, the Governor appointed the Missouri Genetic Advisory Committee (GAC) to advise the DHSS in all genetic programming in the State. In 1988, Missouri successfully applied for federal

funding to expand its targeted newborn hemoglobinopathy program to a universal newborn screening program that began in May 1989. When federal funding ended in 1991, the DHSS identified State funding to maintain that program. In that same year, the DHSS entered into a partnership with the Department of Mental Health to fund the Healthy Children's Initiative that expanded and enhanced genetic services. This included the development of outreach services, improved clinical laboratory support, and increased genetic professional manpower.

As indicated above, Missouri has implemented many of the genetic service components identified in the Guidelines for State Genetic Services developed by the Council of Regional Networks for Genetic Services (CORN). Those components are further described below.

#### **A. Statewide Genetics Program**

The Bureau of Genetics and Healthy Childhood (GHC), located in the Section of Healthy Families and Youth, Division of Community and Public Health (DCPH), houses the genetics program. The bureau consists of 32 full time staff, one of which is designated as the state genetics coordinator. This bureau serves as a central clearinghouse for all genetic issues in the State, through the GAC. The bureau also monitors and evaluates genetic services provided through the genetic contractors in the State.

#### **B. Tertiary Genetic Centers**

The Department of Health and Senior Services contracts with four tertiary genetic referral centers located in Missouri's four university-affiliated medical schools. The staff of the four centers provides genetic diagnostic evaluations and counseling, genetic screening and genetic education, through regularly scheduled genetic clinics, prenatal clinics, and a variety of other specialty genetic disease clinics (metabolic, Down syndrome, etc). These centers offer a complete range of diagnostic and consultative medical services essential for delivery of effective genetic services. Board certified medical geneticists,

genetic counselors, data entry personnel, and cytogeneticists staff these centers. Medical subspecialties available include, but are not limited to: pediatrics, obstetrics, pathology, neurology, and orthopedics.

For the purpose of delivering genetic services, the State is divided into three sections corresponding to the Central, Western, and Eastern portions of the State. The plan for dividing the State is based on available resources, as opposed to existing medical referral patterns. The Eastern section, which includes the City of St. Louis and Southeastern Missouri, is served by Washington University and St. Louis University Medical Centers. Children's Mercy Hospital, which is affiliated with the University of Missouri, provides services in the Kansas City area, while the University of Missouri Hospital and Clinics in Columbia responds to the needs of the rest of the State. These centers are located along Interstate Highway 70, which is the central corridor of the State.

### **C. Outreach Services**

To make medical genetic services available to geographically and/or culturally remote areas of the State, a network of outreach clinics and telehealth medicine clinics in southern Missouri have been established. Outreach clinic services involves sending a genetic team from one of the genetic centers to an identified area for one day every three months. These locations were chosen because each serves as a secondary referral center for a large population area. The telehealth medicine clinics involve utilizing telecommunication technology to enhance the provision of remote health care. Genetic patients receive follow-up genetic examinations, counseling, and health care via telecommunication technology in their community. This reduces the need for families to travel long distances resulting in saving the family both time and money. Telehealth clinics are currently located at these four sites Mt. Vernon, Poplar Bluff, Springfield, and West Plains.

#### **D. Newborn Bloodspot Screening**

State law mandates that all infants born in Missouri be screened for PKU, galactosemia, congenital hypothyroidism, congenital adrenal hyperplasia, and abnormal hemoglobinopathies. In July 2005, an additional six amino acid disorders, eight fatty acid oxidation disorders, and six organic acid disorders were added to the newborn screening panel. In June 2007, CF was added to the newborn screening panel. With the addition of CF to the newborn screening panel, Missouri's screening program screens for 28 of the 29 conditions recommended by the American College of Medical Genetics and the March of Dimes. Screening for biotinidase deficiency is expected to be added in late 2008 and once added Missouri will fully meet the recommendations. When considering secondary disorders detected through newborn screening, newborns born in Missouri are screened for 67 genetic and metabolic disorders.

Each submitting hospital is charged a newborn screening fee of \$25 for each infant tested. This fee was increased to \$50 on August 1, 2005. This fee is used to pay for the expense of the State Public Health Laboratory (SPHL) to perform the newborn screen. The fee is also used to contract with four accredited CF centers to track and follow-up on abnormal immunoreactive trypsinogen (IRT) results. These four accredited CF centers are Cardinal Glennon Children's Medical Center, Children's Mercy Hospital, St. Louis Children's Hospital, and University Hospitals and Clinics.

The SPHL is centrally located in the State and conducts initial tests for each of the 27 disorders. Those infants who are found to be positive on the newborn screen for a particular condition are followed-up to determine if either a repeat newborn screen or a confirmatory test has been done and those infants confirmed to be positive are entered into a system of health care. It is at this point that the Missouri newborn screening program falls short in that it only provides short-term tracking; once the infant is enrolled into a system of

health care the Department's involvement with that child and family end. While the Department can state how many infants were positive and entered into a system of health care, it cannot state what happened to those children, i.e. stay in the health care system, stay on medication, stay on diet, keep health care appointments, moved out of state, etc.

Newborn screening is more than just screening the newborn - it should always be part of a system that includes screening, follow-up, diagnosis, treatment, and evaluation, as necessary. The primary objective of each state's newborn screening system is to ensure that every newborn screened receives appropriate and timely services. Currently, the newborn screening program does not provide long term tracking of these children nor does it contract with a treatment center to provide long-term tracking. A component of this plan is to determine ways in which the DHSS can track these babies long-term. With the DHSS integrated data system, GHC plans to incorporate long-term tracking.

A system that allows the DHSS to partner with healthcare providers is needed to provide long-term tracking of infants who are positive for one of the five conditions.

#### **E. Missouri Newborn Hearing Screening Program (MNHSP)**

Missouri legislation authorized mandatory newborn hearing screening effective January 1, 2002. The purpose of the MNHSP is to assure all babies born in Missouri receive a hearing screen and appropriate follow-up, including audiologic evaluation, enrollment into early intervention, and medical intervention, when indicated. Statistics show that three newborns per 1,000 have some degree of permanent hearing loss. Congenital hearing loss is more common than cleft lip or Down syndrome. Early identification of hearing loss and enrollment in appropriate intervention services during the first six months of life allows children who are deaf or hard of hearing to take

advantage of the critical first few years of life, when language, whether spoken or signed, is acquired and allows children to develop language at a level equal to that of their hearing peers.

Currently, the M NHSP employs two Regional Representatives (RRs) who follow-up each newborn who missed or failed the hearing screen. The RRs contact the parents of newborns who need to return for an initial screen, a rescreen or obtain an audiologic evaluation. Additionally, they contact birth hospitals and primary care physicians to obtain screening results that were not submitted to the DHSS. Their goal is to ensure infants are screened no later than one month after birth, diagnosed by three months of age, and enrolled into early intervention no later than six months of age.

The M NHSP successfully collaborates with numerous entities. The M NHSP works with the Department of Elementary and Secondary Education in order to acquire data about the intervention services children with hearing loss receive through First Steps, Missouri's Part C program. Additionally, Missouri State University provides a contract audiologist consultant who advises the M NHSP in audiological matters and who is available to provide assistance to newborn hearing screening programs throughout the state. All Missouri birth facilities and audiologists report hearing screening and evaluation results to the M NHSP, as required by state statute.

#### **F. Organization of Teratology Information Specialists (OTIS))**

Missouri Teratogen Information Services was a statewide program run by the University Hospitals and Clinics. This program has been discontinued but calls are routed, via a toll free phone number, to OTIS. The mission of OTIS is to stimulate and encourage the dissemination of knowledge, education, and research in the field of clinical teratology, and to improve the abilities of teratology information services to provide accurate and timely information about prenatal exposure with the overall objective of improving the public

health. Such teratogens may include medications used during pregnancy, exposure to viruses during pregnancy, and exposure during pregnancy to various chemicals used both indoors and outdoors. OTIS receives approximately 60 calls per year from Missouri residents.

#### **G. Sickle Cell Anemia Program**

The Sickle Cell Anemia Program provides information to the public and health professionals about sickle cell disease and sickle cell trait, and promotes and provides screening, testing and referral, counseling, and follow-up services for Missouri citizens at-risk for sickle cell conditions. Sickle cell disease affects about 1/400 African Americans in Missouri and 1/12 has sickle cell trait, the carrier state of the disease. The program supports five hemoglobinopathy resource centers throughout the state to ensure that comprehensive medical services are available to individuals and families with sickle cell disease.

Educational programs stressing inheritance and treatment and management of sickle cell disease, are directed at the general public, health professionals, and targeted populations. In addition, information is distributed in the form of pamphlets, brochures, and posters.

All newborns in the State are screened for sickle cell conditions. In 2006, fifty-five (55) infants were identified through newborn screening with sickle cell disease. The purpose of newborn screening is to identify infants with the condition and ensure that appropriate treatment is initiated. Other age groups are also tested, but on a much more targeted basis, i.e., family members of an infant identified with sickle cell trait.

Parents of infants identified with sickle cell trait and other carrier conditions are offered educational genetic counseling. Counseling sessions are non-directive and include information on inheritance. Approximately 1,500

individuals with trait conditions are identified each year through the screening component.

The five hemoglobinopathy resource centers located throughout the State provide expertise in hematology and the treatment of sickle cell disease; conduct follow-up on clinically significant newborn screening results and ensure confirmatory testing is completed; and facilitate patient linkages with medical care. Each resource center is located in a hospital that provides inpatient, outpatient, and emergency care. The resource center staff is available for clinical consultation to providers and patients served outside of the center.

**H. Adult Cystic Fibrosis, Sickle Cell, and Hemophilia Treatment Programs**

These treatment programs provide assistance to individuals 21 years of age and over, who meet the eligibility criteria. Medical eligibility is documented with a written diagnosis. Financial eligibility is at or below 185% of the U.S. Department of Health and Human Services' Poverty Income Guidelines.

These programs provide financial assistance for outpatient and inpatient services, prescription medications, home medical equipment, emergency care, and service coordination. In addition, the Hemophilia Treatment Program provides financial assistance for blood factor products.

Currently, the Hemophilia, Sickle Cell, and Cystic Fibrosis treatment programs offer participants a complete treatment package designed to address the medical needs of affected individuals

**I. Metabolic Formula Program**

The Metabolic Formula Program (MFP) provides dietary formula and an annual examination to medically and financially qualified Missouri residents,

of any age, who have been diagnosed with the diseases listed below by acting as the payer of last resort.

Medical eligibility for the Metabolic Formula Program is documented with a written medical diagnosis of one of the following conditions: phenylketonuria (PKU); maple syrup urine disease (MSUD); glutaric acidemia; homocystinuria; methylmalonic acidemia; citrullinemia; argininosuccinic acidemia; isovaleric acidemia; 3-hydroxy-3-methylglutaryl CoA lyase deficiency (HMG); 3-methylcrotonyl CoA carboxylase deficiency (3MCC), propionic acidemia, long-chain 3 hydroxyacyl CoA dehydrogenase deficiency (LCHAD), very-long-chain acyl-CoA dehydrogenase deficiency (VLCAD), and other diseases approved by the Missouri Genetic Advisory Committee.

Financial eligibility is at or below 185% of the U.S. Department of Health and Human Services' Poverty Income Guidelines. An adjustment of \$10,000 is added to the participant's income to allow for additional expenses for medical food that the program does not provide, since service coordination is available regardless of income.

As of September 17, 2007, the financial eligibility guidelines for the MFP were revised based passage of a new law Chapter 376, Section 376.1219 RSMo. These revisions are:

- Birth through five years-of-age: no financial eligibility guidelines and pre-pay cost requirement for formula.
- Six through eighteen years of age: no financial eligibility guidelines, but there is a pre-pay cost requirement for formula based on the percent of federal poverty level (FPL) calculated from the adjusted gross income and the number of family members. **1)** Below 300% of the FPL, zero cost; **2)** 300% - 399% of the FPL, 25% cost; **3)** 400% - 499% of the FPL, 40% cost; **4)** 500% and over of the FPL, 50% cost.
- Nineteen years of age and older, 185% of the FPL with an adjustment of two to the family size with no requirement to pre-pay for formula.

**J. Birth Defects Registry**

The Registry is a passive, multi-source system that identifies infants with birth defects by monitoring information submitted on the birth certificates, hospital patient abstracts, death certificates, and state program enrollment data. Data are population-based and collected statewide, representing approximately 78,000 resident births annually. Outreach correspondence targets parents who have recently had a child born with spina bifida, a cleft defect, or Down syndrome, to assure they are aware of resources to enhance the development of their child and to provide folic acid education to reduce the risk of birth defect recurrence in future pregnancies.

The DHSS coordinates activities related to the Missouri Folic Acid Advisory Committee. The Committee includes medical providers, public health professionals, and educators, who facilitate timely information exchange regarding birth defects prevention and folic acid education in Missouri. Public outreach to expand folic acid consumption and promote healthy behaviors to reduce the risk of serious birth defects is ongoing. The Department has recently been awarded a March of Dimes Community Grant to conduct “An Ounce of Prevention” curriculum implementation training with University of Missouri Sinclair School of Nursing senior nursing students and central Missouri school nurses and teachers. Senior nursing students have been paired with school nurses and/or educators to teach curriculum programming, which targets middle and high school students with the folic acid and birth defects prevention message, in the classroom.

**K. Missouri Health Strategic Architectures and Information Cooperative (MOHSAIC)**

MOHSAIC is an electronic database, which identifies the functions performed by the DHSS and local public health agencies and the data needed to perform these functions. It also provides the architecture for a statewide information network, which can link public and private health care providers

electronically. It is envisioned that MOHSAIC will eventually be the central data repository for the Department. Newborn hearing screening information and newborn blood spot screening information, as well as other program areas such as immunization and lead screening, are entered and stored in MOHSAIC and plans are to add sickle cell trait counseling in the next few months.

MOHSAIC contains a generic registration component that captures the demographic information required to register a client, no matter the services provided. There is a core of required information such as the client's name, date of birth, race, sex, ethnicity, and client number. Additional information captured in this component includes the client's home, mailing, business or contact addresses, and telephone numbers. It has a number of other features that include an electronic interface with the Department of Social Services Medicaid eligibility and managed care plan enrollment files, the client's primary care provider, health care insurance information, and numerous other client specific information. Program and specific health related information is added as a client is provided public health services.

#### **L. Missouri Genetic Advisory Committee**

The Missouri Genetic Advisory Committee is a governor appointed advisory board, which advises the Department on the provision of genetic services. The Committee meets at least once a year, and insures that genetic programs are responsive to the needs of the entire State and equitably funded. To address specific issues, the Committee has five Standing Committees: the Newborn Screening Standing Committee; the Newborn Hearing Standing Committee; the Cystic Fibrosis Standing Committee; the Sickle Cell Anemia Standing Committee; and the Hemophilia Standing Committee. The Standing Committees meet at least yearly and make recommendations to the Genetic Advisory Committee regarding emerging issues and program direction.

## **IV. Other Genetic Related Services in the Department of Health**

### **A. Special Health Care Needs**

The Bureau of Special Health Care Needs (BSHCN) provides services for children and their families with disabilities, chronic conditions, and birth defects, who meet financial and medical eligibility guidelines. Services focus on early identification of children with special needs and funding for preventive, diagnostic and treatment services. Service coordination is provided by DHSS staff and contracted staff throughout the State.

Many of the conditions covered by BSHCN have either a genetic origin or genetic implications that would benefit from genetic counseling and evaluation services. Referral between BSHCN and the genetic service system is necessary to assure that families are receiving needed services.

### **B. Chronic Disease**

The Section of Chronic Disease Prevention and Nutrition Services directs statewide programs that are designed to combat the major causes of premature death, illness, disability, and medical costs in Missouri such as heart disease, cancer, stroke, diabetes, and arthritis. These programs are primarily conducted through two bureaus and within DCPH: the Bureau of Health Promotion and the Bureau of Cancer and Chronic Disease Control. As more is learned about the genetic implications of common chronic diseases, genetics will play a major role in the development of strategies to prevent and/or lessen the complications associated with these conditions.

### **C. Nutritional Health**

Part of the strategic plan for Missouri is to improve the nutritional health of Missourians. The DCPH recognizes the need to integrate nutrition services for improvement of disease outcomes. Conditions such as Cystic Fibrosis,

Sickle Cell Disease, and PKU require nutrition intervention for successful management of these conditions.

Researchers have found that nutrition counseling is also a vital component of the care of children with genetic conditions. Thus, an objective of the genetic service programs is to collaborate with the nutritional programs and other partners to assist families with the coordination of these services. This will include identifying appropriate diet practices and increase access to nutritional health services.

In addition to improving and protecting the health of these citizens, nutrition intervention can save a significant amount of health care dollars. However, the current challenge is to include nutritional services as part of the multidisciplinary treatment approach.

## **V. Goals and Objectives**

The following goals and objectives are based on the premise that current systems of care must be continuously evaluated and revamped to be responsive to the demands of the “new genetics”. They do not provide a detailed description of how genetic services will be finally implemented in Missouri, but rather provide guidelines for the best ways to guarantee the continuation of quality services in the face of evolving genetic technologies.

### **Goal 1: Develop a statewide surveillance system for genetics by integrating and enhancing existing surveillance systems.**

Objectives:

- Maintain a web-based system for capturing NBS and NBHS data. (On-going)
- Develop and implement NBS and NBHS systems with the capability of providers obtaining client information electronically. (On-going)
- Establish data linkages with genetic counseling and evaluation centers. (On-going)

- Integrate NBS and NBHS information with other child information on immunizations, lead screening, Special Health Care Needs, and WIC participation. (On-going)
- Integrate genetic tertiary care center information and NBS follow-up information. (On-going)
- Develop a statewide surveillance system for genetics by integrating and enhancing surveillance systems. (June 2010)
- Increase the number and type of newborn hearing reports in MOHSAIC. (On-going)
- Integrate newborn hearing data with other child health screening and program data and make the integrated data available to other public health and private providers. (June 2010)
- Integrate the newborn hearing MOHSAIC application with an electronic birth certificate application. (January 2010)
- Create a link between the newborn hearing MOHSAIC application and the communicable disease component of MOHSAIC. (June 2010)
- Implement the MOHEAR project, based upon the current pilot Service Coordination pilot project, on a statewide basis. (2011)
- Assist at least nine hospitals with high lost-to-follow-up-following-refer rates with parent physician's reminders, appointment cards, and scripts for relaying refer results to parents. (2011)
- Conduct a statewide early hearing detection and intervention (EHDI) meeting for Missouri stakeholders. (2011)

**Goal 2: Evaluate and improve the availability and accessibility of genetic services.**

Objectives:

- Evaluate and revise the newborn screening system to ensure the long-term follow-up of affected individuals and their families. (July 1, 2010)
- Conduct annual reviews of all genetic service components. (On-going)
- Create mechanisms to routinely assess evolving genetic issues. (On-going)

- Maintain the genetic service network composed of genetic centers, outreach clinics, and specialty programs for conditions with high prevalence rates. (On-going)
- Maintain a case management information system for NBS and NBHS. (On-going)
- Continue telemedicine activities to outstate locations and expand these activities in other genetic programs. (On-going)
- Facilitate trainings for service coordinators of children with special health care needs, consumers, and genetic health care providers to improve the collaboration between agencies and families. (On-going)
- Maintain an information system to obtain outcome information on children with sickle cell disease. (On-going)
- Monitor compliance of screening and testing protocols and improved standards for treatment therapies. (On-going)

**Goal 3: Increase the awareness of patients, professionals, and the general public of the impact of genetic information on health.**

Objectives:

- Maintain a web site to increase access to resources for NBS and genetic conditions that will serve as a link between the DHSS home page and other genetic information services. (On-going)
- Determine, design, and implement targeted instructional opportunities for audiences for whom the subject is personally or professionally relevant, based upon identified need. (On-going)
- Determine, design, and implement selected informational activities for defined high-risk groups that provide detailed knowledge to impact behavior and prevent complications. (On-going)
- Identify folic acid awareness educational needs in Missouri schools and provide evidence-based curriculum materials to address the need. (On-going)

#### **Goal 4: Establish partnerships to support genetic service systems.**

Objectives:

- Enhance Advisory Committee and Standing Committee memberships to include a wider representation of individuals and specialties, such as a medical ethicist, epidemiologist, genetic counselor, pediatrician, and lawyer with HIPAA expertise. (April 2009)
- Maintain an internal DHSS genetics crosswalk team to ensure that a genetic component is added to other programs in the Department, as needed (e.g. chronic disease, environmental, and nutrition programs). (On-going)
- Establish collaborative partnerships with other agencies to enhance the availability and quality of future genetic services. (On-going)
- Enhance mechanisms to offer public and professional input on the ethical dimensions of any aspect of research, including new initiatives in genetic screening and testing. (On-going)

#### **Goal 5: Establish evidence-based genetic services.**

Objectives:

- Complete a newborn screening pilot program to screen for biotinidase deficiency. (December 2008)
- Analyze the impact cultural and social differences have on utilization of genetic services and develop appropriate interventions. (On-going)

## **VI. Evaluation**

The five goals included in this plan are each followed by a list of objectives with target dates for completion. These tasks have been distributed to the appropriate Bureau of Genetics and Healthy Childhood staff for inclusion in the program work plans. The major form of evaluation for this plan will be determining if the objectives have been met by the specified target dates.

A final form of evaluation will consist of sharing the progress with the Missouri Genetic Advisory Committee and the subsequent standing committees. Input from

these medical and consumer representatives will serve as a vital link to the Bureau's perception of goal attainment.